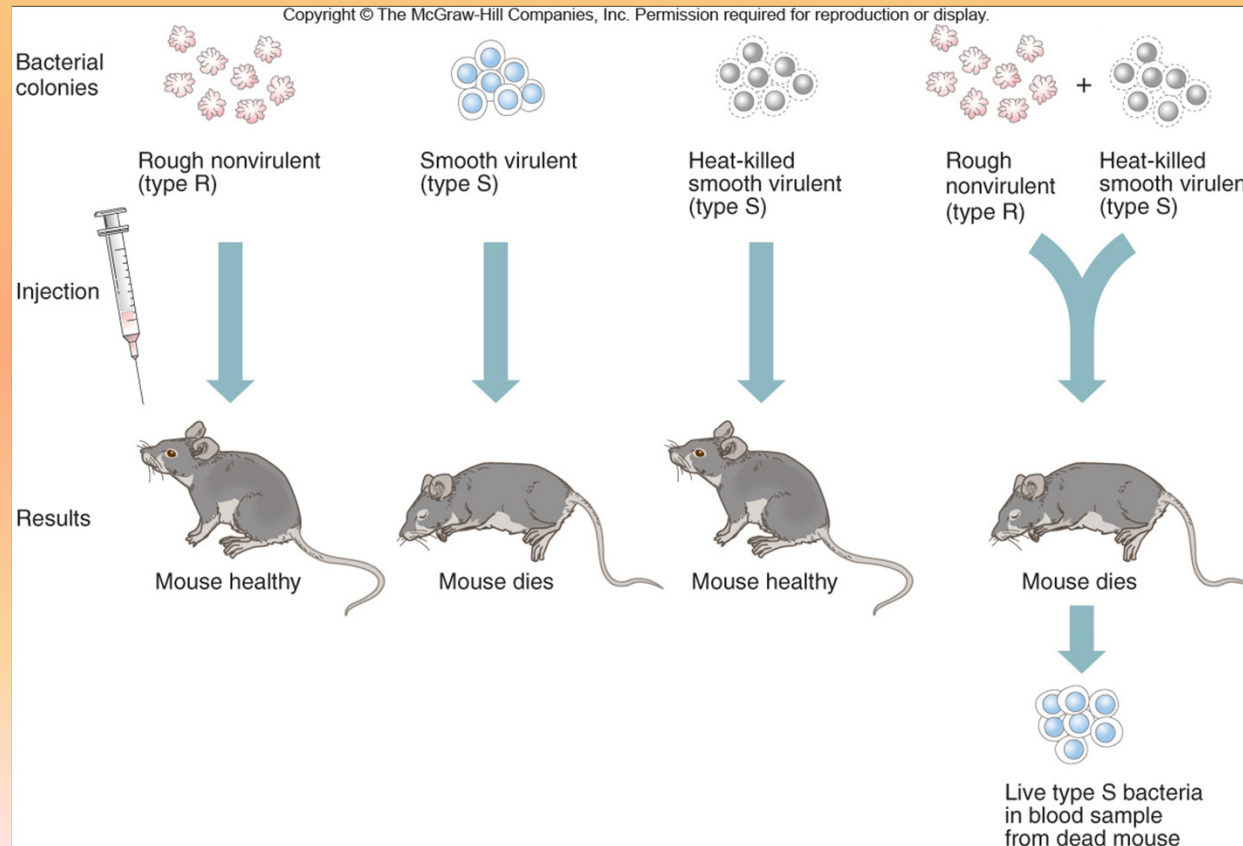


# Identifying the Substance of Genes

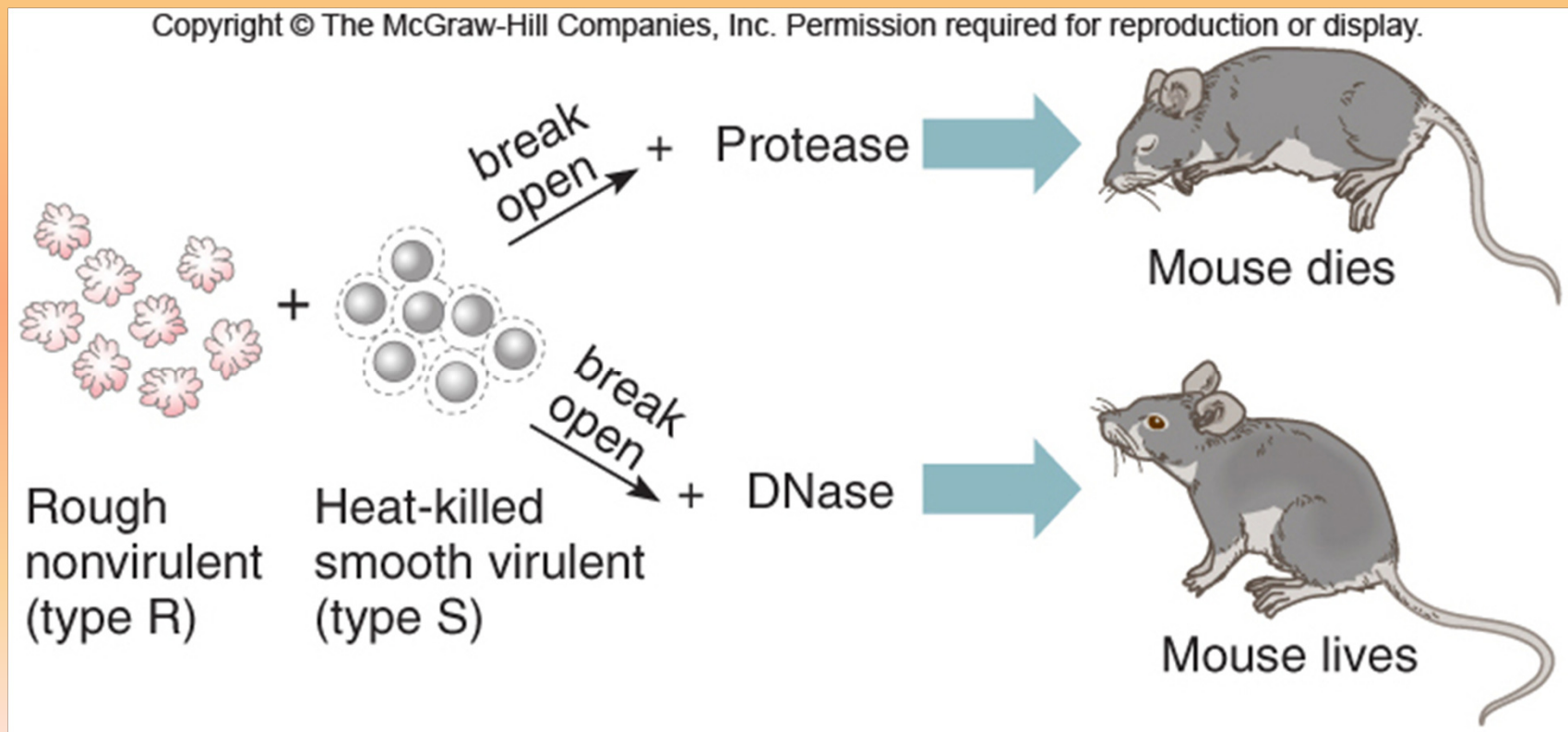
- Griffith

- **Transformation** =

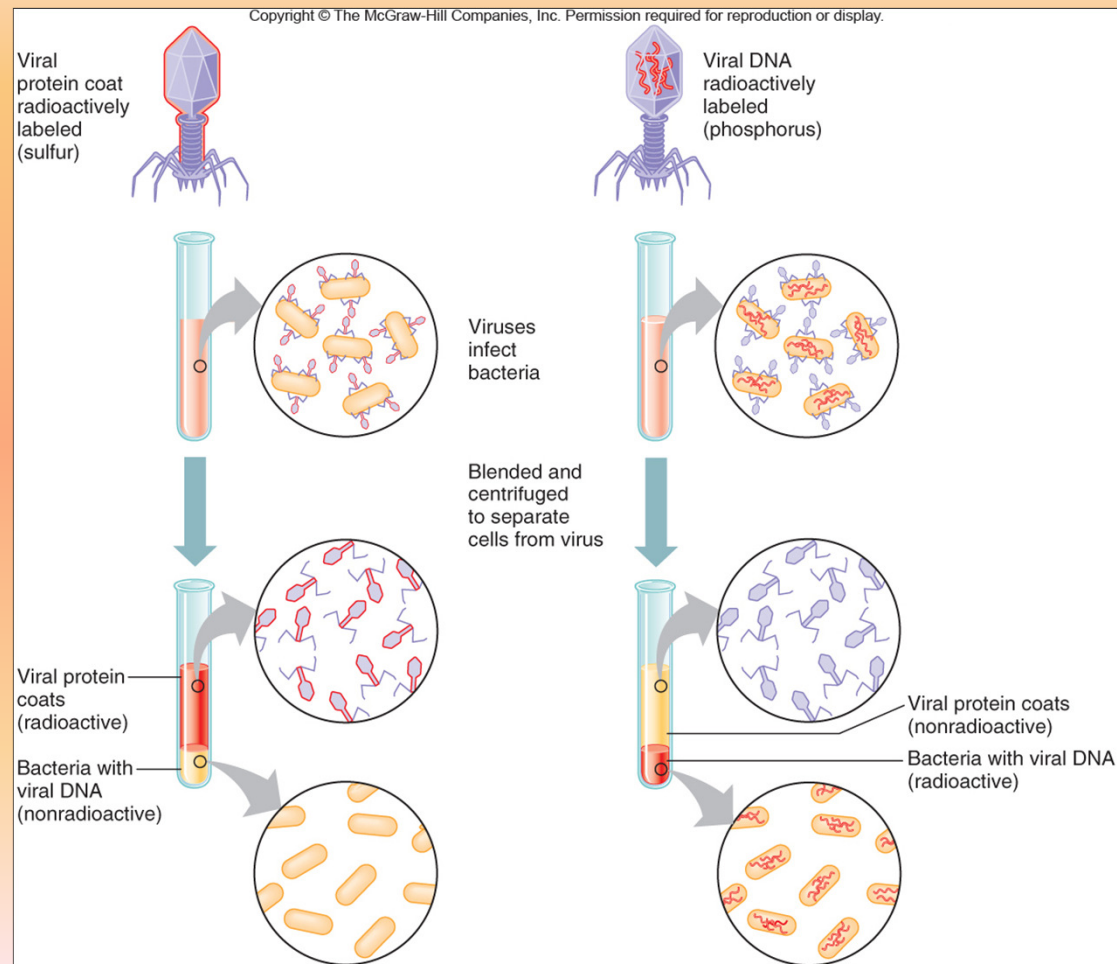
- One strain of bacteria was changed by genes from another strain of bacteria



- Avery, MacLeod, McCarty
  - Determined DNA stores and transmits genetic info from one generation of bacteria to the next



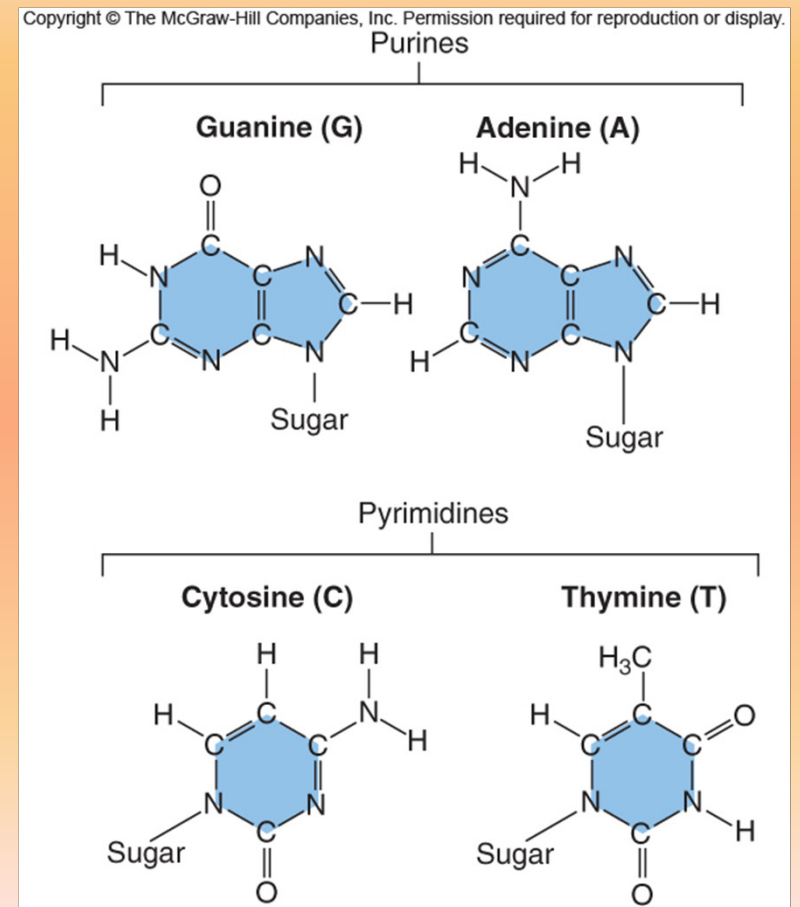
- Hershey and Chase
  - Confirmed DNA (not protein) is the genetic material using radioactive isotopes in viruses



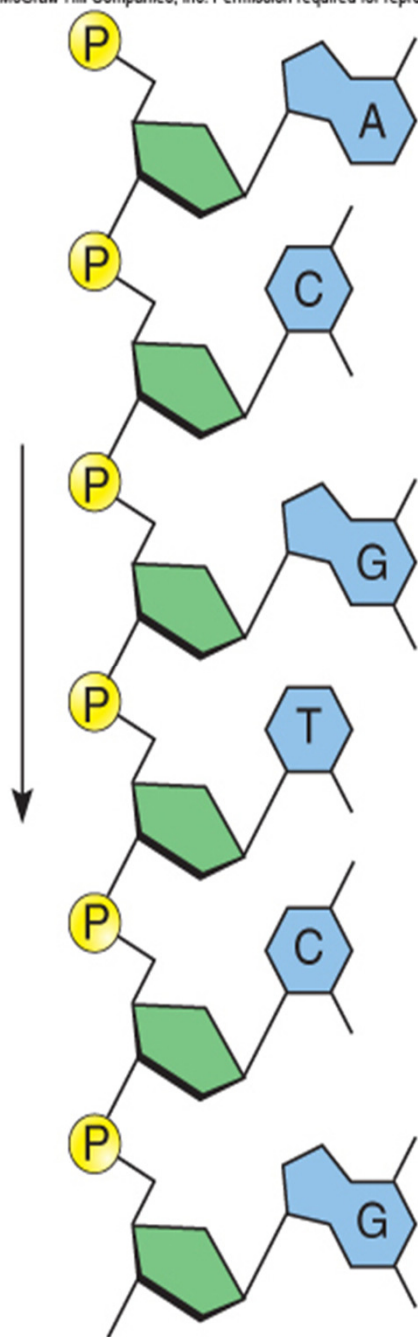
- Conclusions
  - DNA makes up genes
- Assumptions
  - DNA must be capable of
    - Storing information
    - Copying information
    - Transmitting information

# The Structure of DNA

- Building blocks of nucleic acids = **nucleotides**
  - 1 deoxyribose sugar
  - 1 phosphate group
  - 1 nitrogenous base
    - **Purines** (2 ring structure)
      - **Adenine** (A)
      - **Guanine** (G)
    - **Pyrimidines** (single ring)
      - **Cytosine** (C)
      - **Thymine** (T)

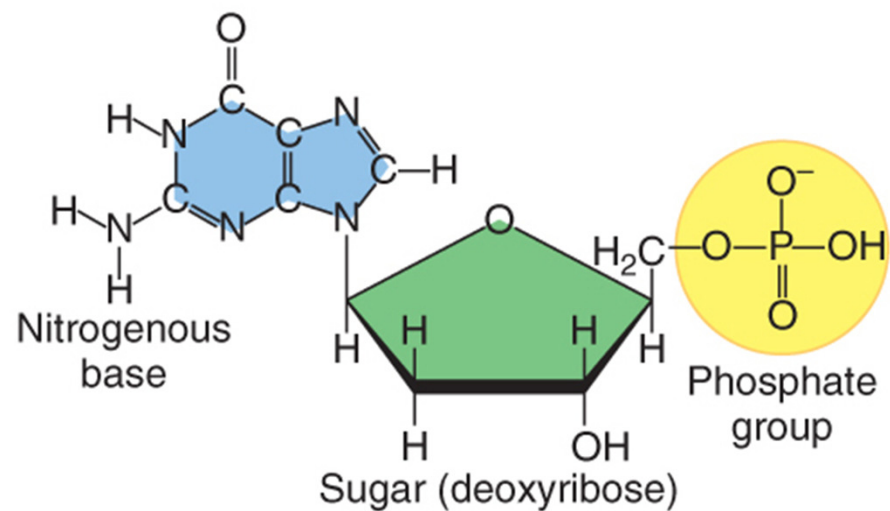


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### Guanine (G)

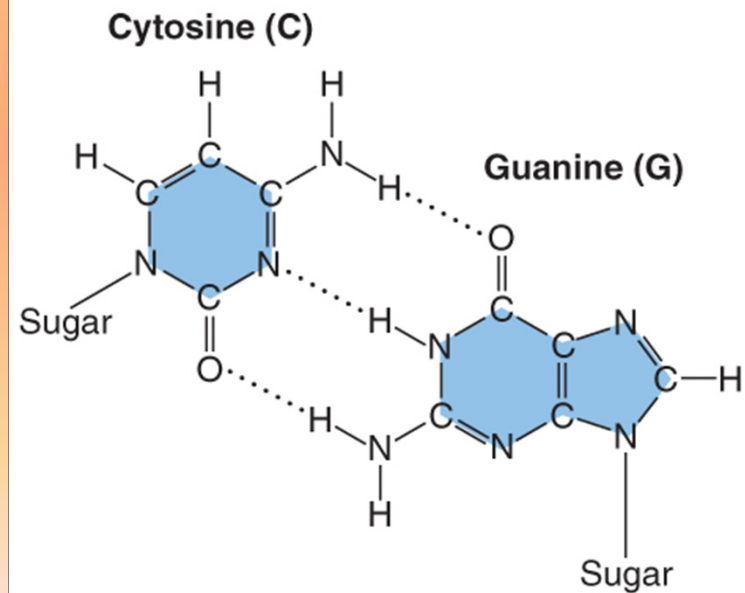
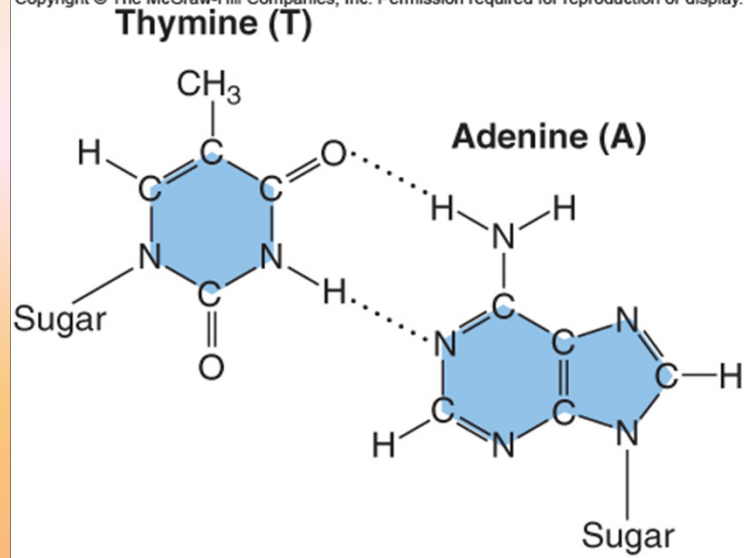




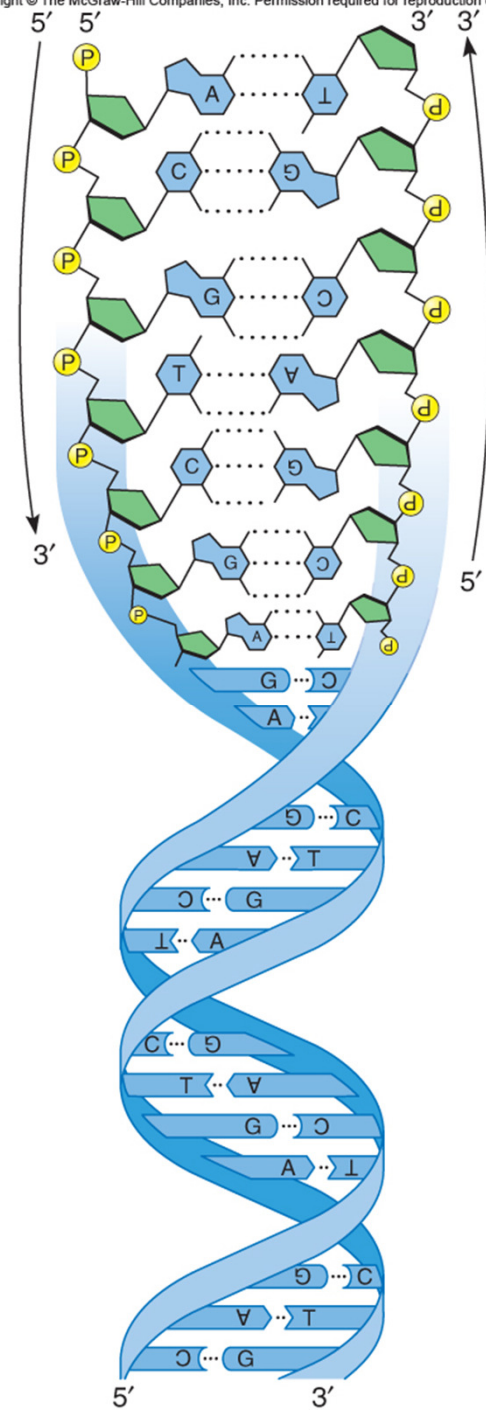
- Chargaff's Rule
  - $A = T$
  - $C = G$
- Wilkins and Franklin
  - Work with X ray diffraction showed patterns indicating DNA's shape is a helix
- Watson and Crick
  - Determined 3-D structure of DNA is a double helix

- Nucleotides join to form long chains
  - Sugar-phosphate backbone
    - Strands are antiparallel
    - 5 prime (5') and 3 prime (3') ends
  - Nitrogenous bases held to those of 2<sup>nd</sup> strand by hydrogen bonds
    - Complementary base pairs
      - **A bonds with T**
      - **C bonds with G**
- Double helix forms when antiparallel, base-paired strands twist

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- **Chromatin** =

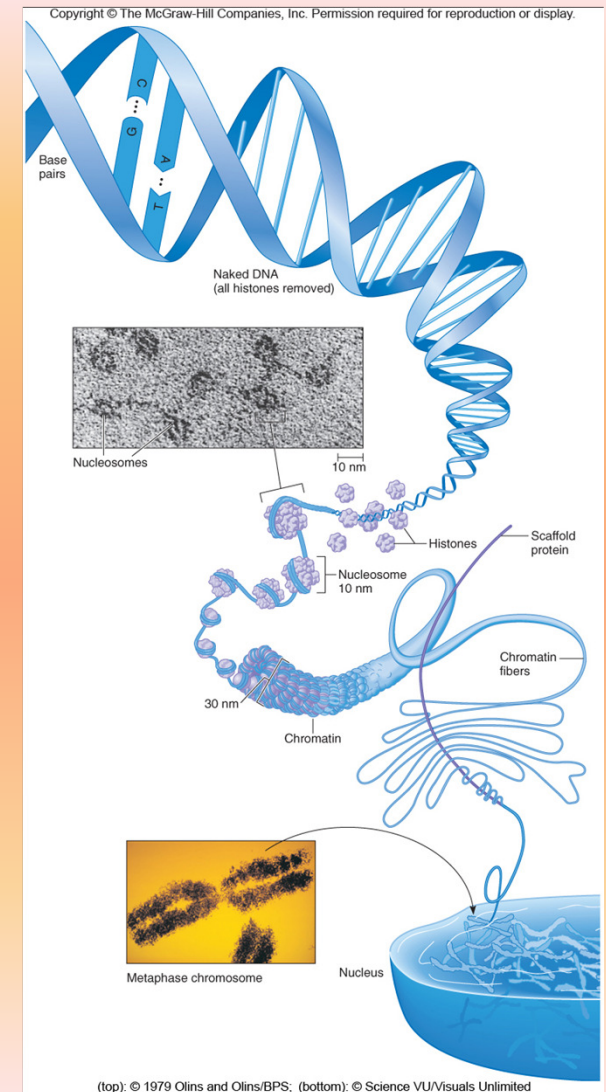
- Complex consisting of DNA tightly coiled around proteins called **histones**

- **Nucleosome** =

- Bead-like unit of chromatin structure

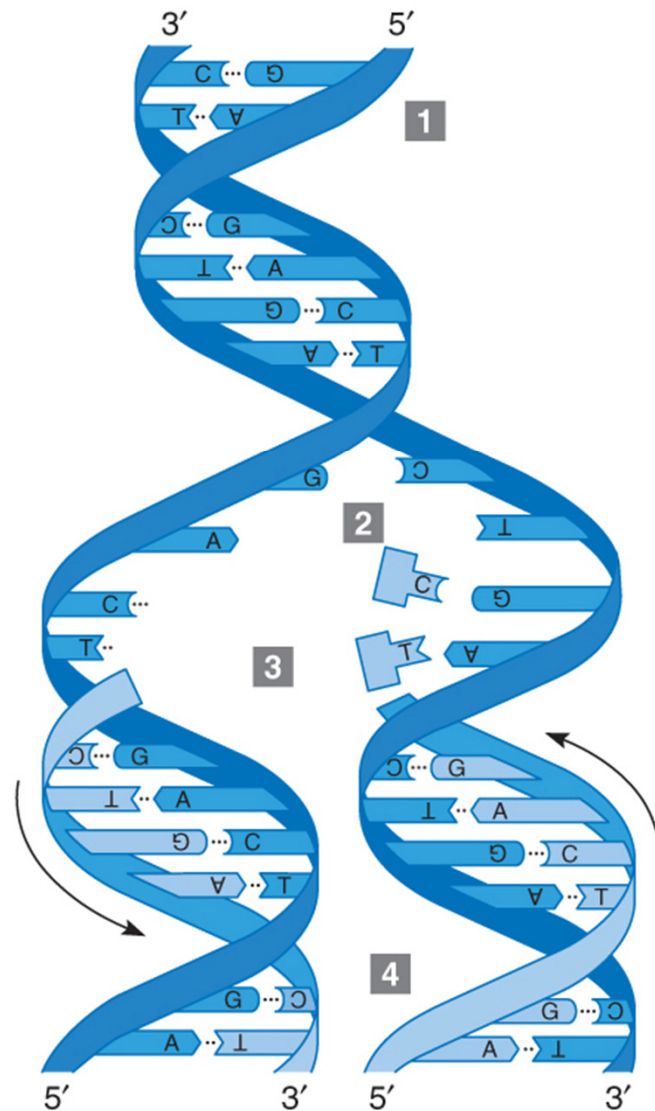
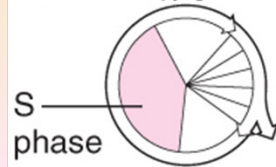
- DNA wraps at several levels, compacting into a chromosome

- Very tightly packed during cell division



# DNA Replication

- **Semiconservative replication** =
  - Each replicated DNA molecule has one original strand and one new strand
- Overview
  1. Strands unwind and separate at several points
  2. **DNA polymerase** adds complementary bases to template
    - **A with T and vice versa**
    - **G with C and vice versa**
  3. Sugar-phosphate backbones seal back up
- Result
  - 2 DNA molecules
  - Identical to each other and original



1 Parent DNA molecule.

2 Parental strands unwind and separate at several points.

3 Each parental strand provides a template for DNA polymerase to bind complementary bases, A with T and G with C.

4 Sugar-phosphate backbones of daughter strands close.

- DNA Replication occurs during the S phase of the cell cycle
- Prokaryotes
  - Replication starts from a single point and proceeds in two directions until finished
- Eukaryotes
  - Replication occurs simultaneously from multiple starting points in both directions



- **Replication fork** =

- Locally opened portions of DNA double helix

- DNA polymerase works directionally

- Only adds new nucleotides to exposed 3' end

- Replication proceeds in a 5' to 3' direction

- Replication on one strand is continuous

- The other is discontinuous

- Produces small pieces called **Okazaki fragments**

- Activities at the replication fork
  1. **Helicase** unwinds and separates strands
  2. **Primase** makes a short stretch of RNA (primer) on the DNA template
  3. **DNA polymerase** adds DNA nucleotides to the RNA primer
    - Also proofreads and corrects mismatched base pairs
  4. **Exonuclease** removes RNA primers
  5. **Ligase** joins Okazaki fragments and seals sugar-phosphate backbone

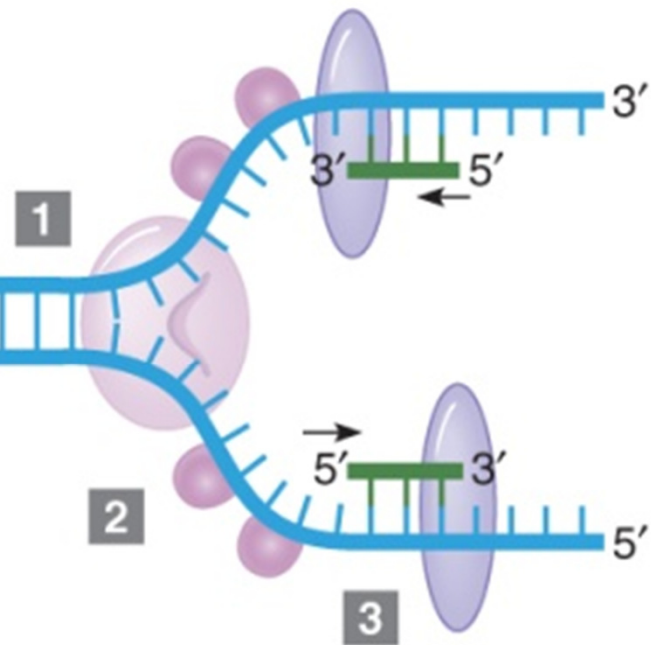
# Activities at the Replication Fork

**1** Helicase binds to origin and separates strands.

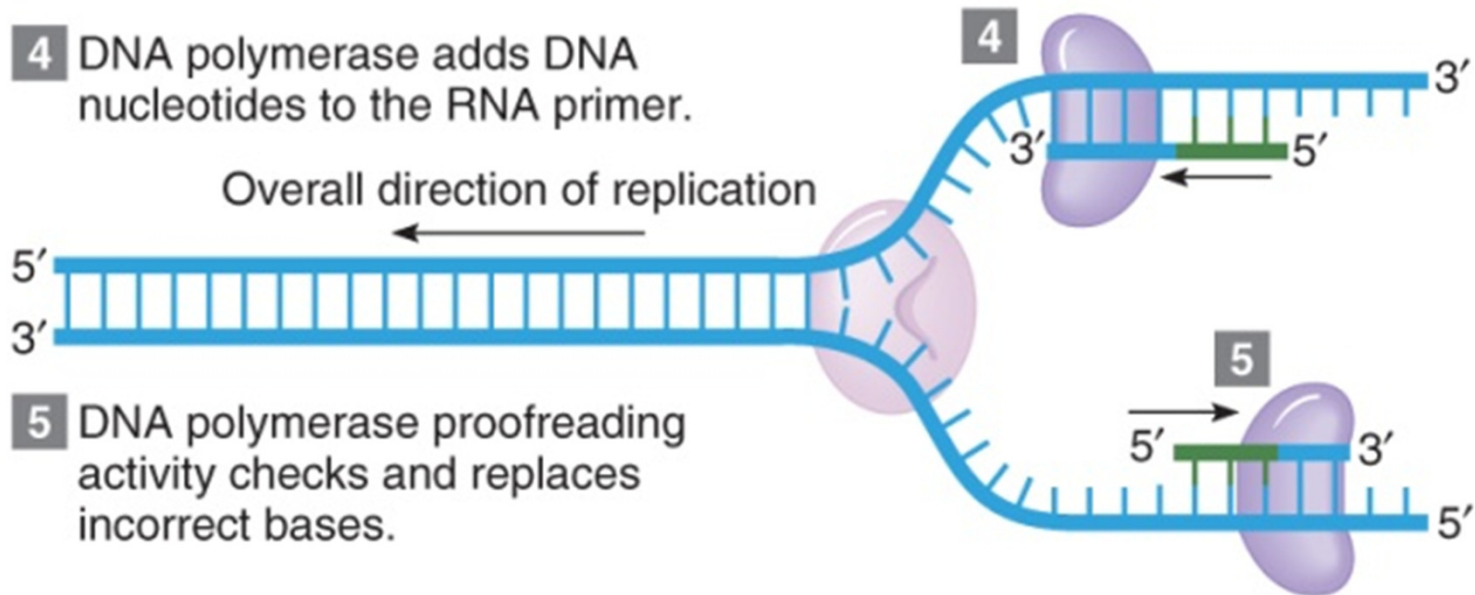
5'  
3'

**2** Binding proteins keep strands apart.

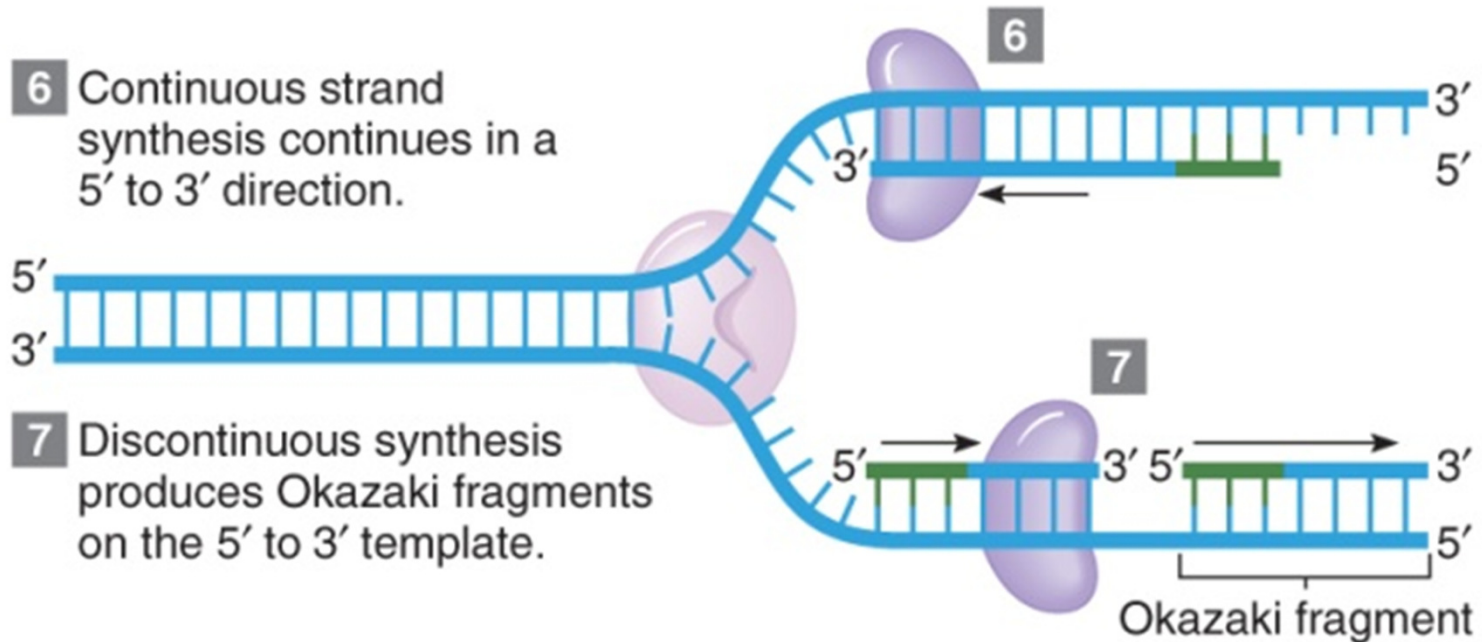
**3** Primase makes a short stretch of RNA on the DNA template.



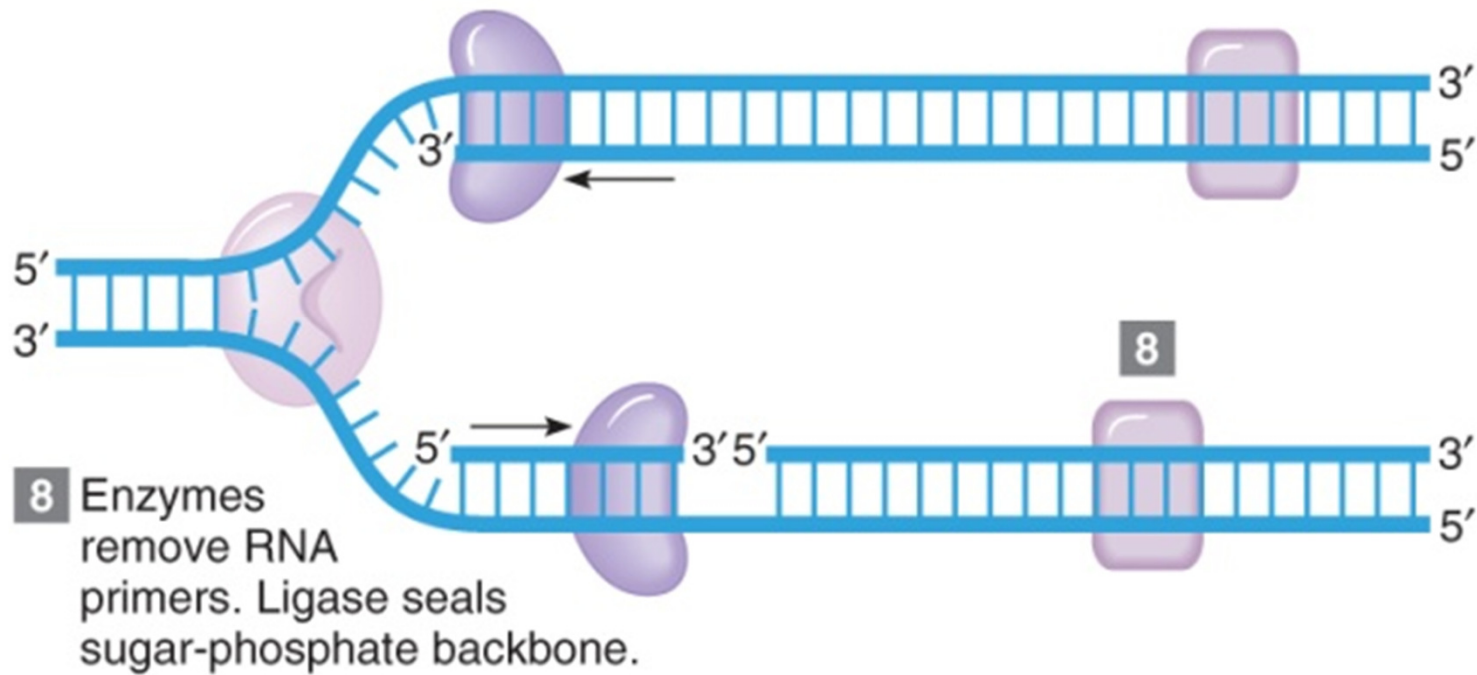
# Activities at the Replication Fork



# Activities at the Replication Fork

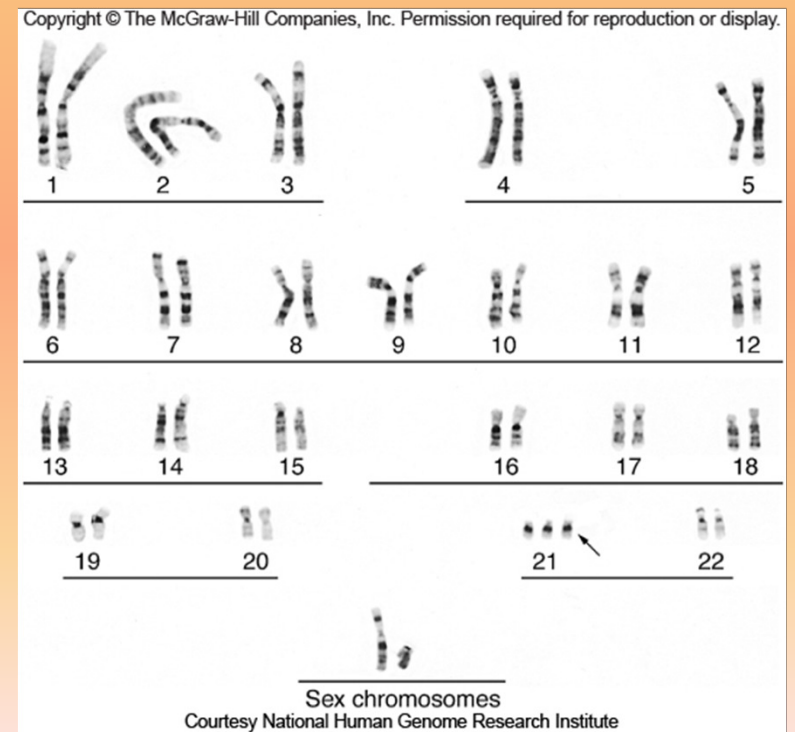


# Activities at the Replication Fork



# Chromosomes

- **Genome** =
  - Entire set of genetic information that an organism carries in its DNA
- **Karyotype** =
  - Shows complete diploid set of chromosomes
  - Grouped in pairs in order of decreasing size
  - Humans have 46 total chromosomes
    - 2 sex chromosomes
      - XX = female
      - XY = male
    - 22 pairs of autosomes





- Centromere position also distinguishes chromosomes

– **Metacentric** =

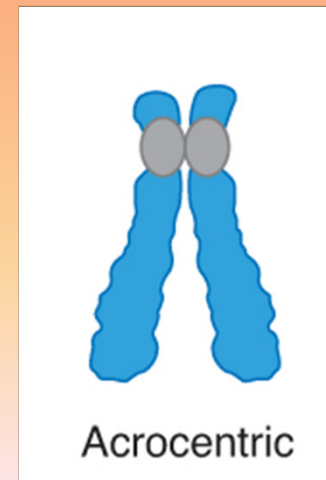
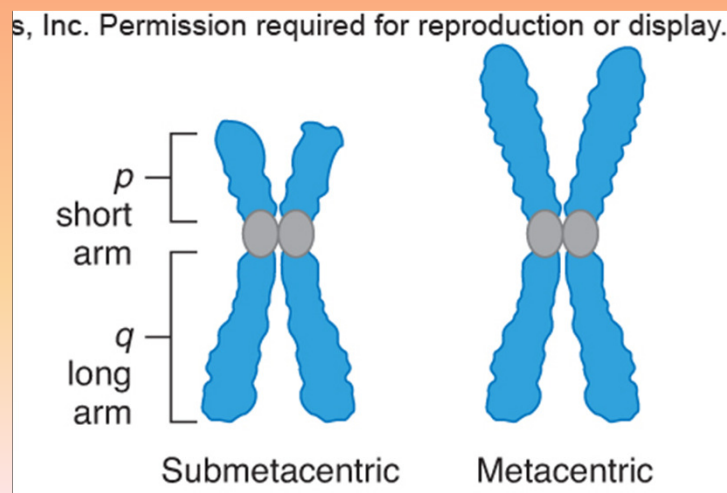
- 2 arms of approx. equal length

– **Submetacentric** =

- One long arm (***q***) and one short arm (***p***)

– **Acrocentric** =

- Pinches off small amount toward one end



# Chromosomal Disorders

# Abnormal Chromosomal Number

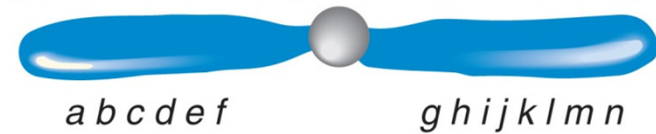
- **Nondisjunction** =
  - Homologous chromosomes do not separate properly
  - Error in anaphase of meiosis
  - Results in abnormal chromosome number
- **Monosomy** =
  - One missing chromosome
- **Trisomy** =
  - One extra chromosome

- Nondisjunction of autosomes could result in:
  - Down syndrome (Trisomy 21)
    - Mental retardation and birth defects
  - Edward syndrome (Trisomy 18)
  - Patau syndrome (Trisomy 13)
- Nondisjunction of sex chromosomes could result in:
  - Turner syndrome (XO)
    - Sterile female
  - Klinefelter syndrome (XXY)
    - Male (typically unable to reproduce)
  - Note: Y with no X won't even be born

# Abnormal Chromosome Composition

- **Deletion** =
  - Loss of part of a chromosome
- **Duplication** =
  - Extra copy of a part of chromosome
- **Inversion** =
  - Reverses direction of parts of chromosome

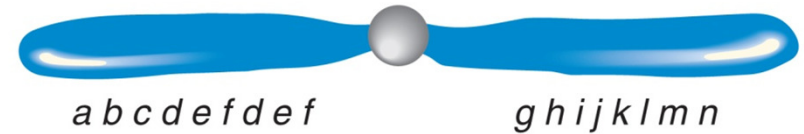
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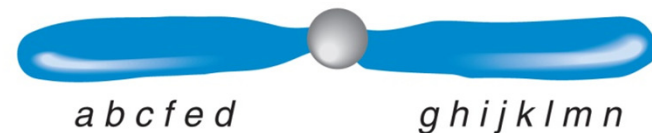
a. Normal sequence of genes



b. Deleted sequence of genes



c. Duplicated sequence of genes



d. Inverted sequence of genes

- **Translocation** =
  - Part of one non-homologous chromosome breaks off and attaches to another
- **Insertion** =
  - Insertion of a larger sequence into a chromosome
  - Due to unequal crossing over during meiosis